

What is Claimed is:

Sub
B10

1. A nucleic acid sequence encoding pyrin.
2. The nucleic acid sequence of claim 1, comprising the coding sequence of **SEQ ID NO: 2** and variations thereof permitted by genetic code degeneracy.

Sub
B11

3. A nucleic acid sequence encoding a familial Mediterranean fever-associated mutant of pyrin.

4. The nucleic acid sequence of claim 3, comprising a mutant pyrin having an amino acid substitution in a rfp (B30.2) domain [**SEQ ID NO: 5**].

5. The nucleic acid sequence of claim 3, encoding mutant pyrin comprising the amino acid sequence of **SEQ ID NO: 7**, **SEQ ID NO: 9**, **SEQ ID NO: 11** or **SEQ ID NO: 13**.

Sub
B12

6. A nucleic acid probe or primer comprising at least fifteen consecutive nucleic acids of *MEFV* [**SEQ ID NO: 1**] or a familial Mediterranean fever-associated mutant thereof.

7. The nucleic acid probe of claim 6, wherein the probe hybridizes to *MEFV* under stringent conditions.

Sub
B13

8. The nucleic acid probe of claim 6, wherein the probe hybridizes mutant *MEFV* under stringent conditions, the mutant *MEFV* comprising a nucleic acid sequence of **SEQ ID NO: 6**, **SEQ ID NO: 8**, **SEQ ID NO: 10** or **SEQ ID NO: 12**.

9. The nucleic acid primer of claim 6, wherein the primer amplifies *MEFV*.

pyrin and correlating hybridization with the presence of wild type or mutant pyrin.

38. The method of claim 36, wherein analyzing comprises sequencing the nucleic acid sequence of pyrin.
39. The method of claim 36, wherein analyzing comprises sequencing or hybridization of a nucleic acid sequence encoding a rfp (B30.2) domain.
40. A method for producing pyrin in a host cell comprising transforming the host cell with a nucleic acid sequence encoding pyrin.
41. The method of claim 40 wherein the host cell is an animal cell.
42. The method of claim 40 wherein the host cell is a mammalian cell.
43. The method of claim 40 wherein the host cell is a human cell.
44. The method of claim 40 wherein the host cell expresses mutant pyrin prior to transformation.
45. A transgenic animal expressing heterologous wild type pyrin or mutant pyrin.
46. A method for screening compounds for use in FMF therapy comprising: administering candidate compounds to the transgenic animal of claim 45.
47. A method for screening compounds for use in inflammatory disease, comprising administering the compounds to the transgenic animal of claim 45.

10. The nucleic acid primer of claim 6, wherein the primer amplifies a nucleic acid sequence encoding a rfp (B30.2) domain of pyrin.
11. An amino acid sequence comprising **SEQ ID NO: 3**.
12. An amino acid sequence encoding a familial Mediterranean fever-associated mutant of pyrin.
13. The amino acid sequence of claim 12, wherein the mutant comprises one or more amino acid substitutions.
14. ~~The amino acid sequence of claim 12, wherein the mutant comprises an amino acid substitution in a rfp (B30.2) domain.~~
15. The amino acid sequence of claim 12, wherein the mutant comprises an amino acid substitution in at least one of amino acid residues 680, 694, 695 or 726.
16. The amino acid sequence of claim 12, wherein the mutant comprises an amino acid substitution corresponding to M680I, M694V, K695R, or V726A.
17. The amino acid sequence of claim 12, wherein the mutant comprises **SEQ ID NO: 7, SEQ ID NO: 9, SEQ ID NO: 11 or SEQ ID NO: 13**.
18. An amino acid sequence encoding pyrin comprising an rfp (B30.2) domain of pyrin [**SEQ ID NO: 5**].
19. The amino acid sequence of claim 18, comprising an amino acid substitution at residue 680, 694, 695, or 726.

00466147 080700

Sub
8/14

20. The amino acid sequence of claim 19, wherein the substitution comprises M680I, M694V, K695R, or V726A.
21. An anti-pyrin antibody that specifically binds wild type pyrin [SEQ ID NO: 3].
22. The antibody of claim 21, wherein the antibody specifically binds to an epitope in a rfp (B30.2) domain.
23. An anti-pyrin antibody which specifically binds familial Mediterranean fever-associated mutant pyrin.
24. The anti-pyrin antibody of claim 23, wherein the antibody specifically binds to a mutation in a rfp (B30.2) domain.
25. The anti-pyrin antibody of claim 23, wherein the antibody specifically binds to pyrin comprising a mutation at residue 680, 694, 695, or 726.
26. The anti-pyrin antibody of claim 23, wherein the antibody specifically binds to mutant pyrin comprising M680I, M694V, K695R, or V726A.
27. The anti-pyrin antibody of claim 23, wherein the antibody specifically binds to mutant pyrin comprising the amino acid sequence of **SEQ ID NO: 7, SEQ ID NO: 9, SEQ ID NO: 11 or SEQ ID NO: 13.**
28. A vector comprising a nucleic acid sequence encoding pyrin [SEQ ID NO: 2] or a familial Mediterranean fever-associated mutant thereof, operably linked to a functional promoter.
29. A host cell transformed with the vector of claim 30.
30. A kit for diagnostic assay comprising:

Sub
8/15

00486147-080700

SUBS
and

a nucleic acid sequence encoding wild-type pyrin; and
at least one nucleic acid sequence encoding a mutant pyrin.

31. A kit for diagnostic assay comprising:
an anti-pyrin antibody which binds wild-type pyrin; and
at least one anti-pyrin antibody which binds mutant pyrin.

5019

32. A kit for diagnostic assay comprising:
at least one pair of primers which amplify a nucleic acid sequence encoding
pyrin.

33. The kit of claim 32, wherein the primers amplify a nucleic acid sequence encoding a rfp (B30.2) domain.

34. A method for diagnosing risk of familial Mediterranean fever (FMF), comprising:
analyzing a patient sample for an amino acid sequence of pyrin; and
correlating detection of mutated amino acid sequence with risk of developing FMF.

35. The method of claim 34, wherein analyzing comprises contacting the sample with an anti-pyrimidine antibody and correlating antibody binding with the presence of pyrimidine in the sample.

Sub 7

36. A method for diagnosing risk of familial Mediterranean fever (FMF), comprising:
analyzing a patient sample for a nucleic acid sequence encoding pyrin; and
correlating detection of mutated nucleic acid sequence with risk of developing FMF.

37. The method of claim 36, wherein analyzing comprises contacting the patient sample with a labeled nucleic acid sequence encoding wild type or mutant

[illegible]